Newborn Screening and Genetic Services:
How can we have a better health impact?

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Inaugural Meeting of the CDC Public Health Genomics Collaboration

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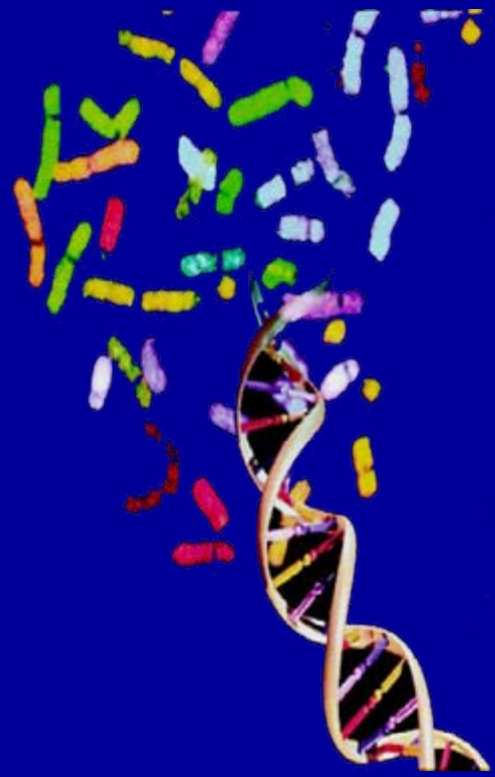

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Overview

- Introduction
  - Genetic services and newborn screening
  - Data for evaluation of health impacts
- Mapping CDC Activities to Life Stage Goals
- Challenges in Quantifying Health Impacts

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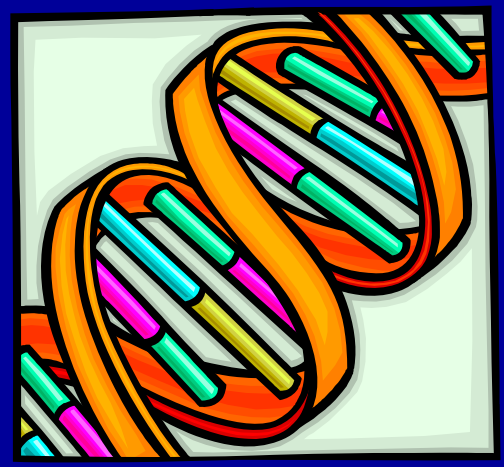
Genetic Services & Screening



- Laboratory testing for genetic disease
  - Newborn screening
  - Carrier screening
  - Diagnostic testing
- Genetic counseling
- Comprehensive treatment for genetic diseases
- Promoting awareness of genetic diseases
  - Family history tools
  - Public and provider education

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Newborn Screening

- Almost all infants screened for
  - Phenylketonuria (PKU)
  - Congenital hypothyroidism (CH)
  - Sickle cell disease (SCD)
  - Galactosemia
- Expanded NBS panels rapidly being adopted
  - Congenital adrenal hyperplasia (CAH)
  - Amino acid disorders (e.g., MSUD)
  - Fatty acid oxidation disorders (e.g., MCAD)
  - Urea cycle disorders
  - Cystic fibrosis (CF)
- Other screening tests on the horizon

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Measures of Health Impact

- 'Hard' metrics
  - Mortality
  - Morbidity
  - Hospitalization rates
  - Disability or functional measures
  - Health care expenditures
- Health-related quality of life of individuals and caregivers
- Multiple measures needed

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Linking Public Health Activities to Health Impacts

- Research on population samples
  - e.g., HFE allele frequencies in NHANES
- Surveillance systems needed to monitor
  - Trends in incidence or prevalence
  - Health outcomes
  - Utilization of services
- Quality assurance
- Intervention research studies
  - Population-based data on outcomes with and without screening and treatment
- Systematic epidemiology reviews
- Cost-effectiveness and cost-benefit analyses

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Evaluation of Outcomes from Newborn Genetic Screening

- 3-state study of children with sickle cell disease detected by NBS
- Workshops and MMWR reports on screening for cystic fibrosis
- Linkage analysis of special education and NBS data in Georgia
- HuGE & systematic reviews
- Economic evaluation of screening for CAH

Quality Assurance in Laboratory Testing

- Newborn Screening Quality Assurance Program (NSQAP)
  - Testing of filter paper matrix
  - External proficiency testing
  - Quality control materials in bulk
  - New technology development
- Laboratory Genomics branch
  - CLIA regulation of genetic tests
  - Quality control materials for genetic testing
  - Facilitate transfer from research labs to CLIA-approved labs
  - Improve reporting of genetic testing results

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Assurance of Quality of Care for Hereditary Bleeding Disorders

- Comprehensive, multidisciplinary care can optimize health outcomes
- 130 hemophilia treatment centers (HTCs) provide care to 15,000 with hemophilia and 10,000 with other bleeding disorders
- CDC study showed that hemophilia patients receiving HTC care have
  - 40% lower mortality risk
  - 40% lower hospitalization for bleeding complications, controlling for confounders
- Current efforts focus on quality of care and quality of life for HTC population

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Single Gene Disorders and Disability

- Collect epidemiologic data on single gene disorders that cause physical and intellectual disability
  - Duchenne/Becker muscular dystrophy
  - Fragile X syndrome
- Assess health outcomes and access to care
  - Mortality
  - Secondary complications
  - Health-related quality of life
- Develop and evaluate interventions for early identification and access to services

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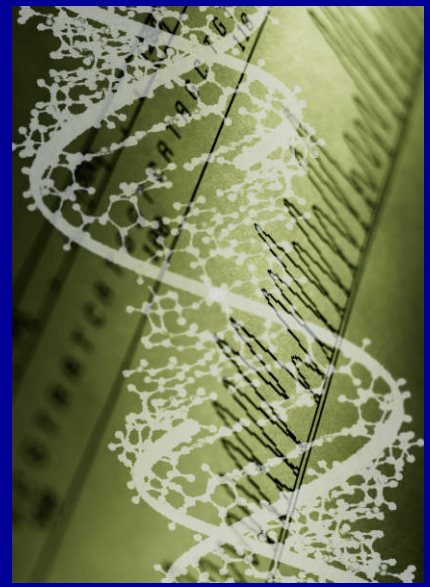
Promoting Awareness of Genetic Diseases

- When population screening is not recommended
- Methods
  - Public awareness
  - Provider education
- Conditions
  - Primary immunodeficiency
    - CDC-Modell Foundation partnership
  - Hereditary hemochromatosis



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CDC Health Protection Goal of Healthy People in Every Stage of Life

- Start Strong: (Infants and Toddlers, ages 0-3 years)
- Grow Safe and Strong: (Children, ages 4-11 years)
- Achieve Healthy Independence: (Adolescents, ages 12-19 years)
- Live a Healthy, Productive, and Satisfying Life: (Adults, ages 20-49 years)
- Live Better, Longer: (Older Adults, ages 50 and over)

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Single Gene Disorders and Life Stages

- Single-gene disorders affect individuals across all life stages
- Many genetic disorders that are treated in pediatric clinics are also common in adults
- Assigning a disorder or intervention to just one life stage can be arbitrary
- Genetic programs at CDC need to start mapping their activities to life stages and expected health impacts – the new budget reality

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Single-Gene Disorders and Life Stages: Examples

- Infants and toddlers
  - Newborn screening
  - Early clinical recognition
- Children
  - Services for Fragile X and Duchenne MD
- Adolescents
  - Hemophilia treatment
- Adults
  - Promoting awareness of hereditary hemochromatosis

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How Can We Optimize Health Outcomes for Single Gene Disorders?

- Example: NBS and sickle cell anemia
  - Deaths < 3 years of age now low
  - Deaths at other ages remain elevated
  - Frequent cause of hospitalization and developmental disabilities
- New clinical and public health strategies are needed to optimize outcomes

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What Is Needed to Optimize Single Gene Activities At CDC?

- Identify CDC priorities
  - Health impact
    - Individual level
    - Population-level
  - Work with stakeholders
  - Be responsive to consumers
- Work on logic model of how CDC activities relate to health impacts
- Develop metrics of health impact
- Modify programs to maximize impacts
- Identify opportunities to establish new activities with external partners

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